



## What are they?

### Points To Remember About Heritable Disorders of Connective Tissue

- There are more than 200 heritable disorders that can affect the tissues between the cells of your body that give tissues form and strength.
- All of these diseases are related to problems in genes that are responsible for building connective tissues.
- Some heritable disorders of connective tissue change the look and growth of skin, bones, joints, heart, blood vessels, lungs, eyes, and ears. Others change how these tissues work.
- Treatments can include regular check-ups with your doctor as well as medicines or nutritional supplements.
- A nutritious diet, exercise, and healthy lifestyle habits can also help.

There are more than 200 heritable disorders of connective tissue that can affect the tissues between the cells of your body that give tissues form and strength. All of these diseases are directly related to problems in genes that are responsible for building connective tissues. The disorders are called “heritable,” because they are passed on from parent to child.

Some heritable disorders of connective tissue change the look and growth of skin, bones, joints, heart, blood vessels, lungs, eyes, and ears. Others change how these tissues work. Many, but not all, are rare.

Common heritable disorders of connective tissue include:

- **Ehlers-Danlos syndrome** mostly affects the skin and joints. Connective tissue becomes weak, which can cause loose joints and fragile, sagging skin.
- **Epidermolysis bullosa** affects the skin, causing blisters.
- **Marfan syndrome** can affect the heart, blood vessels, lungs, eyes, bones, and ligaments.

People with this syndrome may be unusually tall and thin, with long arms and legs.

- **Osteogenesis imperfecta** causes bones to break easily. Sometimes they break for no obvious reason.

## Who gets them?

Heritable disorders of connective tissue can affect anyone. Some of these disorders are seen at birth. Others are seen later in life.

If you have a heritable disorder of connective tissue, it was passed down to you from one or both of your parents.

## What are the symptoms?

Each heritable disorder of connective tissue has its own symptoms. Some examples are:

- **Bone growth problems:** Bones may become brittle, too long, or too short.
- **Joint issues:** Joints may be too loose or too tight.
- **Skin problems:** Skin may be loose, hang in folds, or blister.
- **Blood vessel damage:** Blood vessels may be weak or become blocked.
- **Height issues:** Some types of disorders cause people to be unusually tall or short.
- **Head and facial structural problems:** Certain disorders can make the head and face look different from others.

## Is there a test?

To diagnose heritable disorders of connective tissue, doctors look at:

- Family history.
- Medical history.
- Results from a physical exam.
- Lab tests, which may confirm some disorders.

You may wish to seek genetic counseling if you wish to have a child. A genetic counselor can help you estimate the risk of having a child with the disease. The counselor can also give you information about tests that look for the problem gene in you or your child.

## How are they treated?

Heritable disorders of connective tissue are a wide range of disorders, each requiring a specific treatment. Treatments can include:

- **Regular check-ups** with your doctor to keep track of tissue changes.
- **Medicines or nutritional supplements** such as:
  - Vitamin B<sub>6</sub> to correct a liver enzyme problem.
  - Drugs to slow the widening of blood vessel coming from the heart.
  - Drugs to strengthen brittle bones.

## Living With Them

Maintaining general health is important if you have a heritable disorder of connective tissue. You should talk to your doctor about a plan that includes:

- A nutritious diet.
- Exercise.
- Healthy lifestyle habits.

## For More Info

### **U.S. Food and Drug Administration**

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

### **Centers for Disease Control and Prevention, National Center for Health Statistics**

Website: <https://www.cdc.gov/nchs>

### **American Academy of Orthopaedic Surgeons**

Website: <https://www.aaos.org>

### **American Academy of Dermatology**

Website: <https://www.aad.org>

### **Coalition for Heritable Disorders of Connective Tissue**

Website: <https://www.chdct2.org>

### **Genetic Alliance**

Website: <https://www.geneticalliance.org>

**National Organization for Rare Disorders**

Website: <https://www.rarediseases.org>

**National Society of Genetic Counselors**

Website: <https://www.nsgc.org>

**Dystrophic Epidermolysis Bullosa Research Association of America, Inc.**

Website: <https://www.debra.org>

**Ehlers-Danlos National Foundation**

Website: <https://www.ednf.org>

**National Association for Pseudoxanthoma Elasticum**

Website: <https://www.pxenape.org>

**National Marfan Foundation**

Website: <https://www.marfan.org>

**Osteogenesis Imperfecta Foundation**

Website: <https://www.oif.org>

**PXE International**

Website: <https://www.pxe.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at [NIAMSInfo@mail.nih.gov](mailto:NIAMSInfo@mail.nih.gov).

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)

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[Epidermolysis Ampollosa: Esenciales: hojas informativas de fácil lectura](#)

[Marfan: Esenciales: hojas informativas de fácil lectura](#)

[Osteogénesis Imperfecta: Esenciales: hojas informativas de fácil lectura](#)

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[Heritable Disorders of Connective Tissue, Questions and Answers about](#)

[Marfan Syndrome, Questions and Answers about](#)

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[Epidermolysis Bullosa, Easy-to-Read Fast Facts](#)

[Epidermolysis Bullosa, Questions and Answers](#)

[Osteogenesis Imperfecta Overview](#)

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[Osteogenesis Imperfecta, What People With Osteogenesis Imperfecta Need to Know ...](#)